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(54) Title: ALTERED NUCLEOTIDE SEQUENCE IN CD40 LIGAND PROMOTOR

(57) **Abstract:** This invention relates to alterations in the CD40 ligand (CD40L) promoter, and methods of identifying individuals at risk for a disease or condition in which an elevated expression of CD40L is a factor. Such diseases include autoimmune diseases such as Rheumatoid Arthritis (RA). Nucleic acids encoding for the altered CD40L promoter are provided. These nucleic acids provide diagnostic and therapeutic tools for evaluating and treating or preventing diseases or conditions in which an elevated expression of CD40L is a factor. The invention provides a diagnostic method in which a blood or tissue sample is taken from an individual at risk for RA, nucleic acid is extracted, and the CD40L promoter sequenced. The invention also provides a diagnostic method in which a blood or tissue sample containing cells expressing CD40L is taken from an individual at risk for RA, the transcriptional activity of the CD40L gene is measured, and compared to a control value. The invention further relates to screening for compounds that modulate the activity of an altered CD40L promoter. Such compounds can be used in treating or preventing diseases in which elevated expression of CD40L is a factor.